Evan H Baugh

List of Publications by Year
in descending order

16	808	933447 10	940533 16
papers	citations	h-index	g-index

16	16	16	2106
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Whole exome sequencing reveals potentially pathogenic variants in a small subset of premenopausal women with idiopathic osteoporosis. Bone, 2022, 154, 116253.	2.9	12
2	The benefit of diagnostic whole genome sequencing in schizophrenia and other psychotic disorders. Molecular Psychiatry, 2022, 27, 1435-1447.	7.9	12
3	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	6.2	6
4	Serine biosynthesis defect due to haploinsufficiency of PHGDH causes retinal disease. Nature Metabolism, 2021, 3, 366-377.	11.9	32
5	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
6	Genetic testing in individuals with cerebral palsy. Developmental Medicine and Child Neurology, 2021, 63, 1448-1455.	2.1	19
7	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921.	2.4	5
8	A pathogenic variant in the <scp>SETBP1</scp> hotspot results in a formeâ€fruste <scp>Schinzelâ€“Giedion</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1947-1951.	1.2	11
9	Pre-detection history of extensively drug-resistant tuberculosis in KwaZulu-Natal, South Africa. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 23284-23291.	7.1	23
10	New insights into tardive dyskinesia genetics: Implementation of whole-exome sequencing approach. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2019, 94, 109659.	4.8	9
11	Somatic SLC35A2 variants in the brain are associated with intractable neocortical epilepsy. Annals of Neurology, 2018, 83, 1133-1146.	5.3	95
12	Why are there hotspot mutations in the TP53 gene in human cancers?. Cell Death and Differentiation, 2018, 25, 154-160.	11.2	393
13	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
14	Role of <scp>WNT</scp>10A in failure of tooth development in humans and zebrafish. Molecular Genetics & Genomic Medicine, 2017, 5, 730-741.	1.2	27
15	Robust classification of protein variation using structural modelling and large-scale data integration. Nucleic Acids Research, 2016, 44, 2501-2513.	14.5	52
16	Real-Time PyMOL Visualization for Rosetta and PyRosetta. PLoS ONE, 2011, 6, e21931.	2.5	55